For the genetic analysis we perform High-Depth Exome Sequencing. The bioinformatic analysis was carried out according to what was previously described by Cordoba et al., 2018, resulting in a list of 594,940 variants. Using the multiplatform application of FileMarker relational databases, we prioritized the variants of interest that fulfilled the following criteria: - Genes coding for proteins belonging to the mTOR pathway, - Exonic mutations and - Non Synonymous mutation. A heterozygous variant in RHEB (NM_005614: exon2: c.119A> T: p.E40V) was found (**Figure 1**), a gene encoding a protein with a fundamental role in the activation of the mTOR pathway. The variant that was considered likely-pathogenic for presenting a REVEL of 0.875, being deleterious for the pathogenicity predictors M_CAP / Polyphen2 / SIFT / FATHMM MKL and being in a highly conserved region and fundamental for the functioning of the protein.

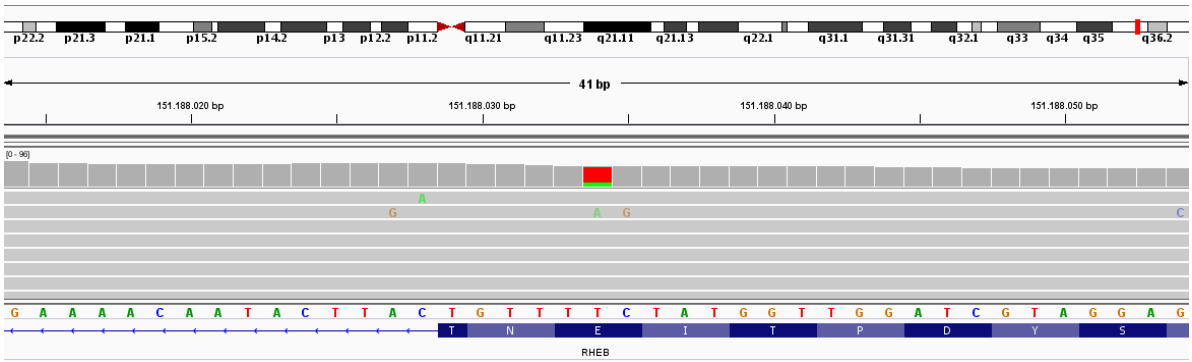

Figure 1.